SUPPLEMENTARY INFORMATION

A common variant of *HMGA2* is associated with adult and childhood height in the general population

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Supplementary Table 1. Basic clinical characteristics of all studies.

		N	% male	Males: age at study (yrs; mean, SD)	Females: age at study (yrs; mean, SD)	Males: average height (cm; mean, SD) or birth length (cm; mean, SD)	Females: average height (cm; mean, SD) or birth length (cm; mean, SD)
a) GWAS	UK WTCCC (T2D	1896	58.2	58.9 (9.9)	57.9 (10.5)	175.4 (7.0)	161.4 (6.6)
	DGI (T2D)	1728	50.6	63.1 (10.3)	65.4 (10.5)	174.3 (6.4)	161.1 (6.2)
	DGI (Controls)	1648	48.6	58.4 (10.5)	59.2 (10.3)	175.6 (6.2)	162.4 (5.9)
b) Replication samples	UKT2D GCC (T2D)	1958	58.1	64.0 (9.1)	64.4 (9.7)	173,7 (6.7)	159.4 (6.2)
	UKT2D GCC (Controls)	1939	51.0	59.6 (11.6)	57.7 (12.2)	176.4 (6.6)	162.5 (6.5)
	ALSPAC mothers (Pop)	6780	0	-	28.4 (4.7)	-	164.0 (6.7)
	EFSOCH parents (Pop)	1856	49.6	32.9 (6.0)	30.4 (5.2)	177.9 (6.6)	165.0 (6.3)
	FINRISK97 (Pop)	6553	46.3	47.9 (13.1)	47.1 (12.7)	175.6 (7.1)	162.3 (6.3)
c) Extreme height samples	European American (5^{th}-10^{th} centile)	1057	48.0	57 (9)	55 (10)	167.1 (1.4)	153.2 (1.5)
	European American (90^{th}-95^{th} centile)	1132	51.9	56 (9)	54 (10)	186.9 (2.0)	172.0 (1.8)
	Polish (5^{th}-10^{th} centile)	512	53.9	55 (10)	56 (9)	164.7 (1.9)	153.4 (1.1)
	Polish (90^{th}-95^{th} centile)	506	47.0	54 (9)	55 (10)	180.9 (1.3)	169.6 (0.9)
d) Children*	ALSPAC children	5100	49.8	11	11	150.2 (7.2)	151.6 (7.2)
e) Birth[†]	EFSOCH children	748	52.7	0	0	50.6 (2.1)	49.8 (2.0)
	ALSPAC children	6079	52.2	0	0	51.2 (2.2)	50.5 (2.2)

*ALSPAC children are offspring of the participants included in the adult study and data are given for the eldest age available (11 years).

[†]ALSPAC birth data are for the same participants as those in the childhood study and EFSOCH children are offspring of the participants included in the adult study. Twins were excluded from birth length analyses in addition to those born before gestation of 36.00 weeks.

Supplementary Table 2: Summary data for rs1042725 in all cohorts.

Study	rs1042725 genotype					
	TT		CT		CC	
a)GWAS	N	Mean (SD)	N	Mean (SD)	N	Mean (SD)
UK WTCCC (T2D)	508	-0.103 (0.956)	924	0.004 (0.979)	464	0.114 (1.059)
DGI (T2D)*	325	-0.054 (0.970)	650	0.017 (0.975)	365	0.159 (0.981)
DGI (Controls)*	225	-0.060 (0.933)	569	0.044 (0.993)	314	0.111 (0.957)
b) Replication samples						
UKT2D GCC (T2D)	477	-0.064 (0.967)	1018	-0.016 (0.990)	463	0.082 (1.016)
UKT2D GCC (Controls)	437	-0.143 (0.923)	997	0.038 (0.992)	505	0.048 (1.067)
ALSPAC mothers (Pop)	1649	-0.063 (0.997)	3354	-0.0005 (0.976)	1777	0.052 (0.991)
EFSOCH parents (Pop)	451	-0.101 (0.988)	925	0.006 (1.025)	480	0.087 (0.953)
FINRISK97 (Pop)	1464	-0.099 (0.973)	3262	0.012 (0.999)	1805	0.032 (0.970)
c) ALSPAC children						
Age 7 years	1438	-0.068 (0.999)	3084	-0.028 (0.979)	1545	0.108 (1.006)
Age 8 years	1208	-0.050 (0.980)	2700	-0.025 (0.996)	1339	0.083 (0.996)
Age 9 years	1280	-0.058 (0.988)	2834	-0.018 (1.003)	1441	0.084 (0.995)
Age 10 years	1238	-0.060 (0.992)	2736	-0.024 (0.998)	1406	0.092 (0.994)
Age 11 years	1173	-0.049 (0.999)	2584	-0.014 (0.994)	1343	0.071 (0.997)

GWAS = genome wide association study. T2D = type 2 diabetes patients. “Controls” indicates participants selected from the general population with exclusion of T2D patients. “Pop” = population-based. *Genotype counts, means, and standard deviations (SD) are given for unrelated DGI individuals only. Means and SD are given in height Z-score units.

Supplementary Table 3: tag SNPs used to dissect the LD block around *HMGA2* rs1042725

SNP rs ID	Chromosome	Physical Position
rs7487625	12	64606263
rs1156095	12	64613521
rs10506473	12	64622005
rs1042725[1]	12	64644614
rs7970350	12	64646431
rs7968902	12	64649337
rs1383303	12	64657648
rs7968682[1]	12	64658147
rs7972653	12	64659166
rs1585897	12	64669587
rs1600576	12	64669838
rs7966895	12	64670110
rs12810075	12	64670469

[1]In total, 13 SNPs were genotyped in the extreme height and FINRISK97 panels. Rs1042725 and rs7968682 were found in the GWAS. They are used for tagging but were not part of the 11 additional genotyped SNPs mentioned in the text.

Supplementary Table 4. Association of sitting height and leg length with rs1042725 genotypes in the ALSPAC study.

Trait and age (yrs)	Gender	Total N	Mean trait value in cm (95% CI) by genotype			Per-C allele effect size (SE)*	*P* value*
			TT	CT	CC		
a) Leg length							
7	Male	3118	57.8 (57.6, 58.1)	57.9 (57.8, 58.1)	58.3 (58.1, 58.5)	0.090 (0.018)	8×10^{-7}
	Female	2946	57.5 (57.3, 57.8)	57.8 (57.6, 57.9)	58.3 (58.0, 58.5)		
9	Male	2790	66.2 (65.9, 66.5)	66.2 (66.0, 66.4)	66.7 (66.4, 67.0)	0.073 (0.019)	9×10^{-5}
	Female	2760	66.0 (65.7, 66.3)	66.2 (66.0, 66.4)	66.7 (66.4, 66.9)		
10	Male	2706	68.4 (68.1, 68.8)	68.5 (68.2, 68.7)	68.9 (68.6, 69.2)	0.079 (0.019)	5×10^{-5}
	Female	2671	68.3 (68.0, 68.6)	68.6 (68.4, 68.8)	69.1 (68.8, 69.4)		
11	Male	2539	72.4 (72.1, 72.8)	72.4 (72.2, 72.6)	72.9 (72.6, 73.3)	0.066 (0.020)	0.0009
	Female	2558	72.5 (72.2, 72.8)	72.8 (72.6, 73.0)	73.1 (72.8, 73.4)		
b) Sitting height							
7	Male	3118	68.3 (68.0, 68.5)	68.3 (68.1, 68.4)	68.6 (68.4, 68.8)	0.068 (0.018)	0.0002
	Female	2946	67.5 (67.3, 67.7)	67.7 (67.5, 67.8)	67.9 (67.7, 68.1)		
9	Male	2790	73.4 (73.2, 73.6)	73.4 (73.3, 73.6)	73.7 (73.5, 73.9)	0.055 (0.019)	0.004
	Female	2760	73.0 (72.7, 73.2)	73.2 (73.0, 73.3)	73.3 (73.1, 73.6)		
10	Male	2706	75.3 (75.1, 75.6)	75.3 (75.1, 75.4)	75.7 (75.4, 75.9)	0.059 (0.019)	0.003
	Female	2671	75.3 (75.1, 75.6)	75.5 (75.3, 75.7)	75.7 (75.5, 76.0)		
11	Male	2539	77.6 (77.3, 77.9)	77.5 (77.4, 77.7)	77.9 (77.6, 78.1)	0.043 (0.020)	0.031
	Female	2558	78.5 (78.2, 78.8)	78.8 (78.6, 79.0)	78.9 (78.6, 79.2)		

**P* values are calculated under an additive model using linear regression corrected for sex. Effect size (regression coefficient) and standard error values are expressed in SD units.

Supplementary Table 5. Association of adult BMI with rs1042725 genotypes in participants used for genome wide association studies and replication samples.

Study	Total N	Mean BMI in kg/m^2 (95% CI) by genotype			Per-C allele effect size (SE)†	*P* value†
		TT	CT	CC		
a) GWAS						
UK WTCCC (T2D)	1895	30.67 (30.20, 31.15)	30.58 (30.23, 30.93)	30.89 (30.40, 31.40)	0.017 (0.031)	0.59
DGI (T2D)*	1330	27.66 (27.01, 28.31)	27.59 (27.03, 28.16)	28.65 (28.03, 29.3)	0.099 (0.040)	0.02
DGI (Controls)*	1118	26.45 (25.81, 27.11)	26.33 (25.78, 26.89)	26.30 (25.47, 27.15)	0.006 (0.040)	0.91
b) Replication samples						
UKT2D GCC (T2D)	1941	30.99 (30.52, 31.47)	30.90 (30.57, 31.22)	31.31 (30.83, 31.80)	0.023 (0.031)	0.45
UKT2D GCC (Controls)	1930	26.34 (25.94, 26.74)	26.25 (25.99, 26.51)	26.13 (25.76, 26.50)	-0.019 (0.033)	0.57
ALSPAC mothers (Pop)	6446	22.54 (22.37, 22.71)	22.73 (22.61, 22.85)	22.82 (22.65, 22.98)	0.039 (0.018)	0.026
EFSOCH parents (Pop)	1770	25.25 (24.89, 25.62)	24.87 (24.62, 25.13)	24.57 (24.23, 24.92)	-0.089 (0.033)	0.007
FINRISK97 (Pop)	5870	26.71 (26.47, 26.97)	26.65 (26.49, 26.80)	26.74 (26.53, 26.95)	0.003 (0.018)	0.85

GWAS = genome wide association study. T2D = type 2 diabetes patients. "Controls" indicates participants selected from the general population with exclusion of T2D patients. "Pop" = population-based. All means and 95% CIs are back-transformed from logged values. * Means and 95% CIs are based on unrelated individuals only. †*P* values are calculated using linear regression under an additive model, corrected for age and sex. Effect size (regression coefficient) and standard error values are expressed in SD units. For the DGI GWAS, *P* values are given for the whole DGI dataset; a genomic control method was applied to control for relatedness. However, the effect size and SE values shown here are for the unrelated component.

Supplementary Table 6. Association of DXA fat mass and BMI with rs1042725 genotypes in the ALSPAC study.

Trait and study subjects	Total N	Mean trait value (95% CI) by genotype			Per-C allele effect size (SE)*	*P* value*
		TT	CT	CC		
a) DXA-measured fat mass (kg)						
ALSPAC children aged 9	5289	7.12 (6.91, 7.35)	7.19 (7.04, 7.34)	7.21 (7.01, 7.43)	0.012 (0.020)	0.55
b) BMI (kg/m^2)						
ALSPAC children aged 7	6058	16.11 (16.01, 16.20)	16.12 (16.05, 16.18)	16.10 (16.01, 16.19)	-0.002 (0.018)	0.90
ALSPAC children aged 8	4960	16.96 (16.84, 17.09)	17.00 (16.92, 17.09)	16.98 (16.86, 17.10)	0.003 (0.020)	0.90
ALSPAC children aged 9	5552	17.43 (17.29, 17.58)	17.52 (17.42, 17.62)	17.44 (17.30, 17.57)	-0.001 (0.019)	0.95
ALSPAC children aged 10	5369	17.94 (17.79, 18.10)	18.01 (17.91, 18.12)	17.93 (17.78, 18.08)	-0.004 (0.019)	0.83
ALSPAC children aged 11	5097	18.69 (18.52, 18.87)	18.83 (18.71, 18.95)	18.70 (18.54, 18.87)	-0.0002 (0.020)	0.99

**P* values are calculated under an additive model using linear regression, corrected for sex. Effect size (regression coefficient) and standard error values are expressed in SD units. All means and 95% CIs are back-transformed from logged values.

Supplementary Methods

Genome Coverage Calculations

To calculate single-point genome coverage for the 364301 autosomal SNPs, we identified HapMap proxies using an r^2 threshold of 0.8 and a MAF > 5%, and then compared this number to the CEU HapMap count of all autosomal SNPs with a MAF > 5%. We used CEU data from HapMap release 21/Phase II in these analyses and SNPs with a MAF < 5% were ignored.

Tag SNP selection (FINRISK97 and GCI Extreme Panel)

All the DNA panels genotyped in this study contain individuals of European ancestry. Therefore, we used genotype data from the CEU panel (Americans of European ancestry) of the phase II HapMap Project Build 21a to determine the patterns of linkage disequilibrium (LD) in *HMGA2* and to select tag SNPs in the LD block containing rs1042725 and rs7968682. The boundaries of the LD block were determined using the "Solid spine" method implemented in Haploview v3.32[1]; the LD block encompasses 64 kb on chromosome 12 (Chr12:64,606,263-64,670,469). We used the software Tagger[2] to choose tag SNPs that capture common genetic variation such that every SNP in the phase II HapMap Build 21a CEU panel with ≥1% allele frequency within this LD block was captured with a pairwise r^2 ≥0.8 by at least one tag SNP or a multimarker predictor (2-3 SNPs). Our final design (after genotyping failure) uses 11 tag SNPs (beside rs1042725 and rs7968682, see Supplementary Table 3) and 20 multimarker predictors to capture the 42 SNPs in this *HMGA2* rs1042725 LD block in CEU at a mean r^2 of 0.87.

References

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Supplementary Note: Membership of WTCCC and DGI

The Membership of the Wellcome Trust Case Control Consortium is as follows:
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